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Case Report
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A Case of Cerulean Cataract

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Abstract

Cerulean Cataracts are a form of congenital cataracts that are characterized by diffuse blue dotted opacifications across the nucleus and cortex of the lens. They Cerulean cataracts are inherited as an autosomal dominant trait. At least four loci for the cerulean cataract phenotype have been mapped – congenital cataract-1 (CCA1; 17q24), CCA2 (22q11.2-q12.2), CCA3 (2q33-q35), and CC4 (16q22-q23). Multiple causative mutations have been identified, including mutations in the beta-B2-crystallin gene (CRYBB2), gamma-D-crystallin gene (CRYGD), V-MAF avian musculoaponeurotic fibrosarcoma oncogene homolog gene (MAF), and the major intrinsic protein of lens fiber gene (MIP). Patients with cerulean cataracts usually have preserved visual acuity and rarely need cataract extraction before adult age. Because many newborns are asymptomatic until 18-24 months of age, cerulean cataracts are considered to be a form of developmental cataract rather than a true congenital cataract. Progression of cerulean cataracts is slow and may not become significant until the third or fourth decade of life, when patients begin to notice a gradual decrease in vision in both eyes. However, children who develop signs of visually significant cataracts such as nystagmus and amblyopia may require cataract surgery earlier. This case report is about a woman who was referred to ophthalmologist as she had changes in retina. She is Georgian and getting diagnosed with cerulean cataract in this country is very rare thus making this patient very important for this geographical region. She has no history of allergy, she has been prescribed reading glasses (+2.5).

Keywords: Cerulean Cataract, Autosomal Dominant, Mutations.

Introduction

A cataract is a term used to describe the opacification of the crystalline lens inside the eye, which impedes the passage of light and represents one of the most Important causes of vision loss [1,2]. Cataract is responsible for ~40-80% of the estimated 45 million cases of blindness That occur across the globe [3].

Cerulean or blue dot cataract is a phenotypic variant of cataract, this phenotype was first described by Vogt6 in 19225, as early onset bilateral cataracts with Concentric layers of bluish-white opacities which may form large cuneiform shapes in the mid- Periphery [4,6]. Cerulean cataracts are inherited as an autosomal dominant trait. At least four loci for the cerulean cataract phenotype have been mapped – congenital cataract (CCA1; 17q24), CCA2 (22q11.2-q12.2), CCA3 (2q33-q35), and CC4 (16q22-q23) [8]. Multiple causative mutations have been identified, including mutations in the beta-B2-crystallin gene (CRYBB2), gamma-D-crystalline gene (CRYGD), V-MAF avian musculoaponeurotic fibro sarcoma oncogene homolog gene (MAF), and the major intrinsic protein of lens fiber gene (MIP) Patients with cerulean cataracts usually have preserved visual acuity and rarely need cataract extraction before adult age [1, 2, 9-11].

Because many newborns are asymptomatic until 18-24 months of age, cerulean cataracts are considered to be a form of developmental cataract rather than a true congenital cataract. Progression of cerulean cataracts is slow and may not become significant until the third or fourth decade of life, when patients begin to notice a gradual decrease in vision in both eyes [12]. However, children who develop signs of visually significant cataracts such as nystagmus and amblyopia may require cataract surgery earlier [13].

Case Presentation

A woman is referred to our ophthalmology clinic from the university clinic with slight difficulty in near vision. She complains of near vision problems she needs to keep the book further away in order to read the text. According to her she started experiencing this problem in vision quite recently. She has a history of cholecystectomy 15 years ago she also complains of a hyperactive bladder (incontinence). She was examined by our clinic in a very comprehensive and a systematic way, her examination included visual acuity pachymetry slit lamp examination tonometry and other essential eye tests Tests results.

Visual acuity measured through Snellen's chart

VOD = 1.0 OR 20/10 VOS = 0.7-0.8 OR 20/30

BCNVA= 40/40 with +2.5D

Tonometry: right eye = 16,15mm left eye = 15, 14 mm

No changes on eyelids, conjunctiva, cornea or anterior chamber No change in iris or pupil Lens showed blue and white opacification scattered in nucleus and periphery Vitreous seemed to be transparent Retina of right eye has healthy fundus with no pathological changes Retina of the left eye showed peripheral degeneration She was only prescribed with lubricants and sent to slit lamp examination The patient was examined using slit lamp to confirm the reason for lens opacification, a clean blue opacification was seen on slit lamp and cataract was confirmed a blue color opacification further led the diagnosis towards cerulean cataract. The bluish opacity in lens upon examination was confirmed to be as cataract and since it was a bluish opacity it was confirmed with cerulean cataract. She needs further Oct exam and few more tests to confirm her retinal changes for which she was referred to our clinic. She was also encouraged to go for genetic screenings for checking the congenital link to her disease.

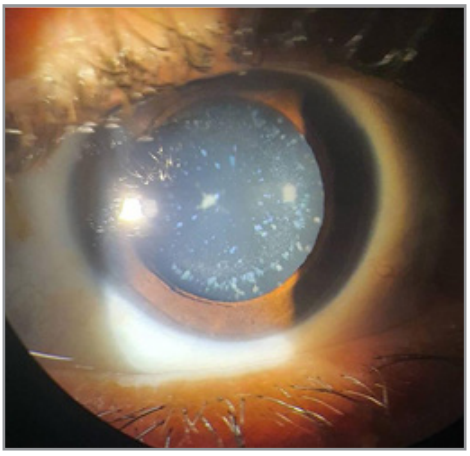

Figure 1: Patient's Right Eye

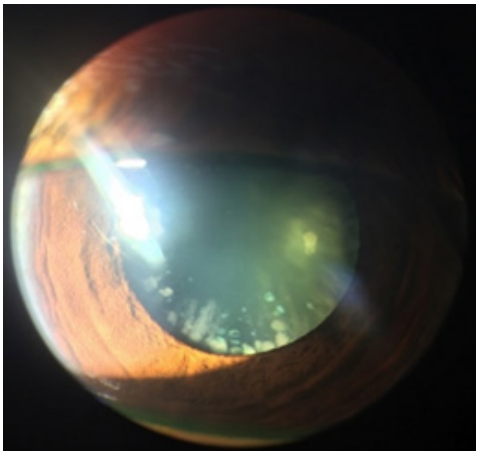

Figure 2: Patient's Left Eye

Discussion

One of the most frequent causes of impaired visual development and permanent visual loss in children is congenital cataract [14,15]. Over a third of all occurrences of congenital cataracts are familial16.

In Western countries, autosomal dominant congenital cataract (ADCC) appears to be the most commonly inherited congenital cataract Cerulean cataracts are a rare form of congenital cataracts also known as blue dot cataracts are a form autosomal dominant[15] congenital cataract that are rare and notable for their unique color and pattern of opacification the lens cortex and nucleus; Cerulean cataracts are associated with mutations in the –crystallins [15,16]. The only component of the 22q -crystallin gene cluster that is significantly expressed in the lens is crystallin B217, the development of cerulean cataract was previously found to originate from missense mutations in the genes that included two b crystallin genes (CRYBB2, CRYBB3) and one pseudogene (CRYBB2P1) [17,18]. (CCA1;17q24), CCA2 (22q11.2-q12.2), CCA3 (2q33-q35), and CC4 (16q22-q23) have all been identified as loci for the cerulean cataract phenotype, latest research indicates that mutations in beta-B2-crystallin gene (CRYBB2), the gamma-D-crystallin gene (CRYGD)19 the V-MAF avian musculoaponeurotic fibrosarcoma oncogene homolog gene (MAF), and the primary intrinsic protein of lens fiber gene (MIP) have all been linked to the formation of cerulean cataract [19]. Modern techniques such as phacoemulsification are used in the case of hard cataracts [20].

Synopsis

The case describes cerulean cataract in a woman, this case is being followed up with genetic tests, the outcome till now is mild cerulean cataract.

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Conclusion

Cerulean cataract is a rare autosomal dominant disorder that develops opaque areas in lens that often have a bluish hue to it may be whitish too. This can be present in childhood but can be diagnosed in adulthood too. This disorder requires more research and investigations related to genetics. The disorder clearly requires more study about the certain genes involved including CRYBB2 CRYGD and MAF genes, which are inherited in an autosomal dominant manner. The disease poses it rarity in the geographical REGION of Georgia and needs proper awareness amongst doctor and patients in order to detect the disease early and identify the genetic link and prevent its adulthood manifestations. Role of parents are important in consulting their children with an ophthalmologist to prevent this disorder and also to diagnose any case of congenital cataract. Genetic screening might work as a bridge to determine this disease if it's being run into families and prevent it in further generations. Awareness regarding this disease would be very beneficial to diagnose it early and prevent adulthood onset

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